The California Expanded AFP Screening Program

Basic booklet for women under 35 years of age

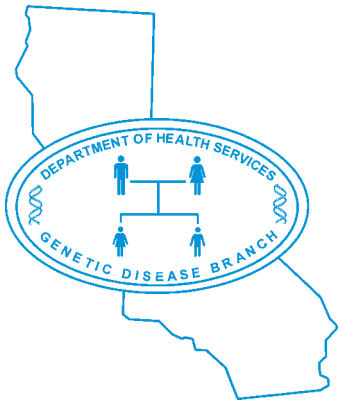
There is a different booklet for ***women 35 years and older.*** These women should ask their doctor or clinic for the green booklet called: **"Choices in Prenatal Testing for Women 35 Years and Older."**

BASIC + HIPAA

California Department of Health Services—Genetic Disease Branch

The California Expanded AFP Screening Program is voluntary. Women can refuse testing without losing insurance benefits, or eligibility or services from State programs.

California law prohibits the use of these results by insurance companies or employers to discriminate against an individual. If you believe that you have experienced discrimination as a result of participation write to the Chief of the Genetic Disease Branch, at the address below.



CALIFORNIA DEPARTMENT OF HEALTH SERVICES
Genetic Disease Branch
850 Marina Bay Parkway, F175
Richmond, CA 94804
866-718-7915 *toll free*
www.dhs.ca.gov/gdb
March, 2007

The California Expanded AFP Screening Program

Every pregnant woman wonders about the health of her fetus (unborn baby) and the possibility of birth defects. The Expanded AFP blood test can help detect some birth defects. This booklet describes the test **for women under 35 years of age at delivery**. It is a woman's own decision whether to have the test or not. A consent/refusal form is at the end of this booklet.

The Program helps detect open neural tube defects, abdominal wall defects, Down syndrome, trisomy 18, and Smith-Lemli-Opitz syndrome. (These birth defects are described on pages 5 and 6.) The Expanded AFP Screening Program consists of: the Expanded AFP blood test first, followed by diagnostic tests if needed.

The test results apply only to this pregnancy.

Who should consider having the Expanded AFP blood test?

All pregnant women.

This blood test is the best way to find out if this pregnancy has a high or low risk of certain birth defects.

If a woman (or the baby's father) has a medical or family history of inherited conditions, she should discuss the test with her doctor. A woman with a high risk pregnancy should also talk to her doctor. There may be special tests that should be done for these women. Some women may need genetic counseling before deciding about this test.

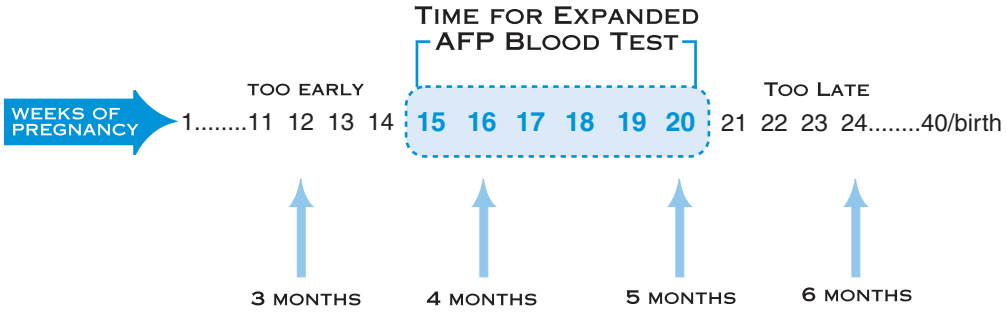
What does the blood screening test involve?

A small amount of blood is taken from the pregnant woman's arm. Her blood is tested for the amount of **AFP** (alpha-fetoprotein), **HCG** (human chorionic gonadotropin), and **UE** (unconjugated estriol). These substances are made by the mother's placenta and the fetus. At each week of pregnancy there are different amounts of these substances in the mother's blood. (What she eats does not affect these substances.)

When is the blood screening test done?

The blood test can only be done reliably **between 15 and 20 weeks of pregnancy**. *The best time is 16 to 17 weeks.* It is important to know how far along the pregnancy is. Ultrasound is very useful for this purpose.

The result of the blood test is sent to the patient's doctor or clinic within 1-2 weeks.



What does a "screen negative" result mean?

It means that the risk for ***certain*** birth defects is low enough that the Program does not consider follow-up tests necessary. The risk is calculated by measuring the amounts of AFP, HCG and UE in the woman's blood and also by considering her age. About 90% of women tested will have a "screen negative" result.

Since the blood test is just a screening test, ***there is still a chance that the fetus may have a problem*** — even when the result is "screen negative."

What does a "screen positive" result mean?

It means that the risk for certain birth defects in this pregnancy is higher than usual (including neural tube defects, abdominal wall defects, Down syndrome, trisomy 18, and Smith-Lemli-Opitz syndrome). The risk is calculated using the amounts of AFP, HCG and UE found in the woman's blood. Her age is part of the calculation of risk for some defects.

Most of the time, however, the reason for this result is ***not*** a birth defect. The **most common reasons** for a "screen positive" result include:

- the due date is earlier or later than thought *or*
- there is more than one fetus (twins, triplets) *or*
- the substances in the blood varied more than usual, without any known pregnancy problem.

To determine the reason for the "screen positive" result, **genetic counseling and follow-up diagnostic tests are offered** and paid for by the Program. Since receiving this result may cause anxiety, it is important to remember that ***most women with "screen positive" results will have normal follow-up tests and healthy babies.***

Summary of Test Results

"screen negative" →	No follow-up tests are offered by the Program.
"screen positive" →	Follow-up tests are provided at no extra cost at a State-approved Prenatal Diagnosis Center.

If the test is "screen positive," what happens then?

A woman with a "screen positive" result will be **called by her doctor or clinic**. She will be offered diagnostic services at a **State-approved Prenatal Diagnosis Center**. When authorized, these are the follow-up services covered by the Program:

- **Genetic counseling** - A professional counselor discusses the pregnancy and family medical history. Questions are answered to help the woman make decisions about further testing.
- **Ultrasound** - A picture of the fetus is made using sound waves. This picture shows the age of the fetus and whether there are twins. The detailed ultrasound done at a Prenatal Diagnosis Center can also detect certain birth defects.
- **Amniocentesis** - A small amount of fluid is taken out of the uterus by experienced, State-approved doctors. The fluid and the fetal cells in it are tested for specific birth defects.

Women may refuse any of these services at any time.

Please remember: most women with "screen positive" results will have normal follow-up tests and healthy babies.

What if the follow-up tests show that the fetus has a birth defect?

Information will be given to the woman by a doctor or genetic counselor at the Prenatal Diagnosis Center. They will discuss the type of birth defect that has been found and any available treatments. They will also discuss options for continuing or ending the pregnancy. The woman can then make a decision.

The Expanded AFP Screening Program does not pay for any other medical services after the follow-up tests. Referrals for special support services are available.

Birth Defects Found by the Program

What birth defects may be found through follow-up testing?

Open neural tube defects, abdominal wall defects, Down syndrome, trisomy 18, Smith-Lemli-Opitz syndrome, and some other birth defects may be found.

Neural Tube Defects (NTDs)

As a fetus is forming, the neural tube extends from the top of the head to the end of the spine. This develops into the baby's brain and spinal cord. The neural tube is completely formed by 5 weeks after conception.

If there is an opening in the spine, it is called **spina bifida**. This defect often causes paralysis of the legs. It may also cause loss of bowel and bladder control. Frequently, there is water-on-the-brain (hydrocephaly) which requires surgery.

Anencephaly occurs when most of the brain does not develop. This defect causes the death of the fetus or newborn.

Abdominal Wall Defects

Fetuses with these defects have **abnormal openings** on the abdomen. Intestines and other organs form outside the body. Surgery after birth often corrects the defect.

Birth Defects Found by the Program

Down Syndrome

Down syndrome is a common cause of mental retardation. Heart defects are often present, as well. Down syndrome is caused by an extra chromosome #21. Chromosomes are packages of genetic material found in every cell of the body. Birth defects can occur when there are too few or too many chromosomes.

Down syndrome can occur in the fetus of a woman of any age. However, as a woman gets older, her chances increase for carrying a fetus with Down syndrome.

Trisomy 18

Trisomy 18 is caused by an extra chromosome #18. Babies with trisomy 18 have severe mental retardation and physical defects. They usually die before birth or in early infancy.

Smith-Lemli-Opitz Syndrome

This is a very rare birth defect. Babies with Smith-Lemli-Opitz syndrome cannot make cholesterol normally. Babies need to make cholesterol, even before birth, to help their brains and bodies develop. Children born with this condition are mentally retarded and may have numerous physical defects.

Birth Defects Found by the Program

How Many Birth Defects Are Found?

These birth defects **do not occur very often**. However, ***if there is one of these birth defects***, the Expanded AFP Screening Program helps detect it. Among all women who have the Expanded AFP blood test and follow-up tests, the Program finds about:

- 97% of the cases of anencephaly
- 80% of the cases of open spina bifida
- 85% of the cases of abdominal wall defects
- 57% of the cases of Down syndrome in the pregnancies of women under 35 years of age
- 55% of the cases of trisomy 18 in the pregnancies of women under 35 years of age
- 60% of the cases of Smith-Lemli-Opitz syndrome

Can the Expanded AFP Screening Program detect every type of birth defect?

No. There are birth defects which **cannot** be detected by Expanded AFP Screening. Even when the blood test is "screen negative," there is still a chance the fetus may have a problem.

How much does the Expanded AFP Screening Program cost?

At this printing, the fee is **$162**, but check with the doctor or clinic about the most current fee. The fee covers the blood test *and* authorized follow-up services at a State-approved Prenatal Diagnosis Center.

The Program mails a bill and insurance form to women who have the blood test. Women with private insurance should complete the insurance form and return it. **In most cases, health insurance companies and HMOs are required to cover the costs of the Expanded AFP testing after you pay any deductible or copay. There is an exception made for self insured employers. Contact your health insurance provider to determine your plan copay.**

Women with Medi-Cal usually do not receive a bill. If they do, they should return the bill with their Medi-Cal number. Women without health insurance may make monthly payments and are responsible for the whole amount.

Any charges for drawing blood are not included in the program fee.

If you have questions about the test or the cost, ask your doctor. After you have decided, please sign the consent/refusal form on the next page.

TEAR HERE AND FILE IN PATIENT'S CHART

CLINICIAN'S COPY
(Remove and file in patient's chart.)

Patient's name_____
(PLEASE PRINT)

ID#_____

CONSENT/REFUSAL

FOR THE CALIFORNIA EXPANDED AFP SCREENING PROGRAM

1. I have read the information about the **California Expanded AFP Screening Program** which is contained in this booklet (or have had it read to me by _____).
2. I have been informed that:
   a) the purpose of the California Expanded AFP Screening Program is to detect most fetuses with open neural tube defects, abdominal wall defects, Down syndrome, trisomy 18, and Smith-Lemli-Opitz syndrome. However, not all such defects can be detected by the Program.
   b) there are other birth defects that cannot be detected by this Program.
   c) if the result is "screen positive," I will need to make a decision regarding follow-up testing. Authorized follow-up tests are covered by the Program and will be discussed with me in more detail.
   d) if the result is "screen negative," the Program will not pay for any follow-up testing.
   e) if the fetus is found to have a birth defect, the decision to continue or terminate the pregnancy will be entirely mine.
   f) participation in the California Expanded AFP Screening Program is voluntary. I can refuse any tests at any time.

(over)

3. I have read the detection rates for certain birth defects as they are described in this booklet.
4 I have been informed that a blood specimen for the California Expanded AFP Screening Program is only reliable between 15 and 20 weeks of pregnancy.
5. I have had my questions answered to my satisfaction.

YES	I request that blood be drawn for the Expanded AFP Screening Program. Signed_____ Date_____ I should have my blood drawn between _____ and _____ month day year month day year
NO	I request that blood <u>not</u> be drawn for the Expanded AFP Screening Program. Signed_____ Date_____

I understand that the blood specimen and information obtained during the testing process become the property of the California Department of Health Services. They may be used for program evaluation or research by the Department or Department-approved scientific researchers without identifying the person or persons from whom these results were obtained, unless I specifically prohibit such use in writing. All information procured by the Department of Health Services, or by any other person, agency or organization acting jointly with the Department in connection with such special studies, shall be confidential. I may obtain additional information about the study or prohibit the use of my specimen by writing to, the Genetic Disease Branch, 850 Marina Bay Parkway, F175, Richmond, CA 94804.

If new information becomes available about a birth defect detected during this pregnancy, the information may be sent to me unless I specifically prohibit it by writing to the Chief of the Genetic Disease Branch at the above address.

TEAR HERE AND FILE IN PATIENT'S CHART

PATIENT'S COPY

Patient's name_____
(PLEASE PRINT)

ID#_____

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NOTICE OF INFORMATION PRACTICES AND PRIVACY PRACTICES CALIFORNIA DEPARTMENT OF HEALTH SERVICES

GENETIC DISEASE BRANCH PRENATAL SCREENING PROGRAM -

EFFECTIVE DATE APRIL 14, 2003

THIS NOTICE DESCRIBES HOW PERSONAL (INCLUDING MEDICAL) INFORMATION ABOUT YOU OR YOUR NEWBORN MAY BE USED AND DISCLOSED AND HOW YOU CAN GET ACCESS TO THIS INFORMATION. PLEASE REVIEW IT CAREFULLY.

Department's Legal Duty. Federal and State laws restrict the use, maintenance and disclosure of personal (including medical) information obtained by a State agency, and require certain notices to individuals whose information is maintained. State laws include the California Information Practices Act (Civil Code 1798 et seq.), Government Code Section 11015.5 and Health and Safety Code Section 124980. The federal law is the Health Insurance Portability and Accountability Act of 1996 (HIPAA), 42 USC 1320d-2(a)(2), and its regulations in Title 45 Code of Federal Regulations Sections 160.100 et seq. In compliance with these laws, you and those providing information are notified of the following:

Department Authority and Purpose for the Prenatal Screening Program. The Department of Health Services collects personal and medical information as permitted in Health and Safety Code Sections 124997, 124980, 125000, 125050, 125055, and 123055. The information is collected and used according to procedures in State regulations (17 CCR 6527, 6529, 6531 and 6532). It is used to estimate the risk of serious birth defects in the pregnancy and provide diagnostic testing for pregnant women.

Continued

If not provided, problems could result such as not detecting an affected fetus, falsely reporting increased risk causing unnecessary invasive testing, or not being able to bill properly for the services provided. This information is collected electronically and includes such things as your name, address, testing results, and medical care given to you.

Uses and Disclosure of Health Information. The Department of Health Services uses health information about you or your newborn for screening, to provide health care services, to obtain payment for screening, for administrative purposes, and to evaluate the quality of care that you or your newborn receive. Some of this information is retained for as long as 21 years. The information will not be sold.

The law also allows the Department to use or give out information we have about you or your newborn for the following reasons:

- For research studies that have been approved by an institutional review board and meet all federal and state privacy law requirements, such as research related to preventing disease.
- For medical research without identification of the person from whom the information was obtained, unless you specifically request in writing that your information not be used by contacting the person listed below.
- To organizations which help us in our operations, such as by collecting fees. If we do, we will make sure that they protect the privacy of information we share with them as required by federal and state law.

The information is otherwise confidential and will not be released without your written authorization. If you choose to sign an authorization to disclose information, you can later revoke that authorization to stop any future uses and disclosures by contacting the Chief of the Genetic Disease Branch.

Continued

The Department may change its policies at any time subject to applicable laws and regulations. If it does so, we will notify you and you may request a copy of our current policies or obtain more information about our privacy practices, by contacting the person listed below or consulting our website at www.dhs.ca.gov/pcfh/gdb. You may also request a paper copy of this Notice.

Individual Rights and Access to Information. You have the right to look at or receive a copy of your or your newborn's health information. If you request copies, we will charge you $0.10 (10 cents) for each page. You also have the right to receive a list of instances where we have disclosed health information about you or your newborn for reasons other than screening, payment or related administrative purposes. If you believe that information in your or your newborn's record is incorrect or if important information is missing, you have the right to request that we correct the existing information or add the missing information. You have the right to ask us to contact you at a different address, post office or telephone number. We will accept reasonable requests.

You may request in writing that we restrict disclosure of your or your newborn's information for health care treatment, payment and administrative purposes. We may not be able to agree to your request.

Complaints. If you believe that we have not protected your or your newborn's privacy or have violated any of your or your newborn's rights and wish to complain, please call or write us at: Privacy Officer, CA Department of Health Services, P.O. Box 942732, Sacramento, CA 94234-7320, (916) 255-5259 or (877) 735-2929 TTY/TDD

Continued

You may file a complaint by calling or writing the **Privacy Officer**, CA Department of Health Services, at the address and telephone number above. You may also contact the Secretary of the Department of Health and Human Services, Office for Civil Rights at 50 United Nations Plaza, Room 322, San Francisco, CA, 94102, telephone (800) 368-1019. Or you may call the U.S. Office of Civil Rights at 866-OCR-PRIV (866-627-7748) or 866-788-4989 TTY.

The Department cannot take away your health care benefits or do anything to hurt you in any way if you choose to file a complaint or use any of the privacy rights in this Notice.

Department Contact – Who Maintains the Information. The information on this form is maintained by the Department of Health Services, Genetic Disease Branch. The Chief of the Genetic Disease Branch may be reached at, 850 Marina Bay Parkway, Richmond, California, 94804, (510) 412-1499. He is responsible for the system of records and shall, upon request, inform you about the location of your records and respond to any requests you may have about information in those records.

AMERICANS WITH DISABILITIES ACT (ADA)

Notice of Information and Access Statement
Policy of Nondiscrimination on the Basis of Disability and Equal Employment Opportunity Statement

The California Department of Health Services (CDHS) complies with all state and federal laws, which prohibit discrimination in employment and provide admission and access to its programs or activities.

The Deputy Director, Office of Civil Rights (OCR), CDHS has been designated to coordinate and carry out the department's compliance with nondiscrimination requirements. Title II of the ADA addresses nondiscrimination and access issues regarding disabilities. To obtain information concerning the CDHS EEO Policies or the provisions of the ADA and the rights provided, you may contact the CDHS OCR by phone at 916-440-7370, TTY 916-440-7399 or write to:

OCR, CA Dept. of Health Services
MS0009, P.O. Box 997413
Sacramento, CA 95899-7413

Upon request, this document will be made available in Braille, high contrast, large print, audiocassette or electronic format. To obtain a copy in one of these alternate formats, call or write:

Chief, Prenatal Screening Section
850 Marina Bay Pkwy, F175 Mail Stop 8200
Richmond, CA 94804
Phone: 510-412-1456
Relay Operator 711/1-800-735-2929

THE CALIFORNIA NEWBORN SCREENING TEST

Newborn screening can save your baby's life or prevent serious brain damage. Newborn screening can identify babies with certain diseases so that treatment can be started right away. Early identification and treatment can prevent mental retardation and/or life-threatening illness.

What Types of Diseases are Screened for in California?

To protect the health of all its newborns, California state law requires that your baby must have the Newborn Screening (NBS) Test before leaving the hospital. The test screens for specific diseases in the following groups:

Metabolic diseases - affect the body's ability to use certain parts of food growth, energy and repair.

Endocrine diseases - babies make too much or too little of certain hormones that affect body functions.

Hemoglobin diseases - affect the type and amount of hemoglobin in red blood cells, often leading to anemia and other problems.

How is the Test Done and Who Pays for it?

A few drops of blood taken from the baby's heel are put on special filter paper. Medi-Cal, health plans, and most private insurance will pay for the test. The cost is included in the hospital bill.

Make Sure You Get This Booklet!

Make sure you get the booklet Important Information for Parents About the Newborn Screening Test from your prenatal care provider or go to our website at www.dhs.ca.gov/gdb then click on Newborn Screening Section for more information about the Newborn Screening Program.